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(54) **PROTEOMIC SCREENING FOR LYSOSOMAL STORAGE DISEASES**

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(57) **ABSTRACT**

Early detection of lysosomal storage diseases (LSDs) including Mucopolysaccharidosis Type I (MPS I) and Pompe Disease can greatly improve patient outcome as each disease can be fatal once symptoms emerge. Screening for MPS I and Pompe Disease using biological samples including dried blood spots (DBS), buccal swab, peripheral blood mononuclear cells (PBMCs), or white blood cells (WBCs) is described. The disclosed methods and assays provide a robust way to screen newborns for LSDs. The disclosed methods and assays can also allow rapid prediction of whether a patient with LSD will develop an immune response to enzyme replacement therapy (ERT), thus improving treatment for patients with LSDs. The disclosed methods and assays can also further reduce the number of false positives caused by pseudo deficiency cases of LSD, such as MPS I and Pompe Disease.

Specification includes a Sequence Listing.